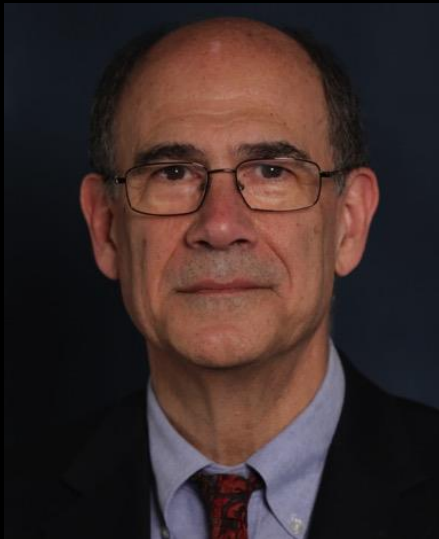




9th Annual Meeting: Wrap-Up

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Importance of Diagnostic Criteria



Research

JAMA Otolaryngology–Head & Neck Surgery | [Original Investigation](#)

Consensus-Based Attributes for Identifying Patients With Spasmodic Dysphonia and Other Voice Disorders

Christy L. Ludlow, PhD; Rickie Domangue, PhD; Dinesh Sharma, PhD; H. A. Jinnah, MD, PhD; Joel S. Perlmutter, MD; Gerald Berke, MD, PhD; Christine Sapienza, PhD; Marshall E. Smith, MD; Joel H. Blumin, MD; Carrie E. Kalata, MS; Karen Blindauer, MD; Michael Johns, MD; Edie Hapner, PhD; Archie Harmon, PhD; Randal Paniello, MD; Charles H. Adler, MD, PhD; Lisa Crujido, MS; David G. Lott, MD; Stephen F. Bansberg, MD; Nicholas Barone, PhD; Teresa Drulia, PhD; Glenn Stebbins, PhD



Importance of Diagnostic Criteria



Neurology
2013

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Development and validation of a clinical guideline for diagnosing blepharospasm



ABSTRACT

Objective: To design and validate a clinical diagnostic guideline for aiding physicians in confirming or refuting suspected blepharospasm.

Methods: The guideline was developed and validated in a 3-step procedure: 1) identification of clinical items related to the phenomenology of blepharospasm, 2) assessment of the relevance of each item to the diagnosis of blepharospasm, and 3) evaluation of the reliability and diagnostic sensitivity/specificity of the selected clinical items.

Results: Of 19 clinical items initially identified, 7 were admitted by content validity analysis to further assessment. Both neurologists and ophthalmologists achieved satisfactory interobserver agreement for all 7 items, including "involuntary eyelid narrowing/closure due to orbicularis oculi spasms," "bilateral spasms," "synchronous spasms," "stereotyped spasm pattern," "sensory trick," "inability to voluntarily suppress the spasms," and "blink count at rest." Each selected item yielded unsatisfactory results in discriminating patients with blepharospasm from healthy subjects and patients with other eye disorders. Combining the selected items, however, improved diagnostic sensitivity. The best combination, yielding 93% sensitivity and 90% specificity, was an algorithm starting with the item "stereotyped, bilateral, and synchronous orbicularis oculi spasms inducing a sense of eye strain or fatigue" and followed by recognition of "sensory trick" or, alternatively, "increased blink count at rest."

Conclusion: This study provides an accurate and valid clinical guideline for diagnosing blepharospasm. Use of this guideline would make it easier for providers to recognize and manage blepharospasm in clinical and research settings. *Neurology*® 2013;81:236-240



Importance of Natural History



Movement disorders

JNNP 2019



ORIGINAL RESEARCH

Risk of spread in adult-onset isolated focal dystonia: a prospective international cohort study

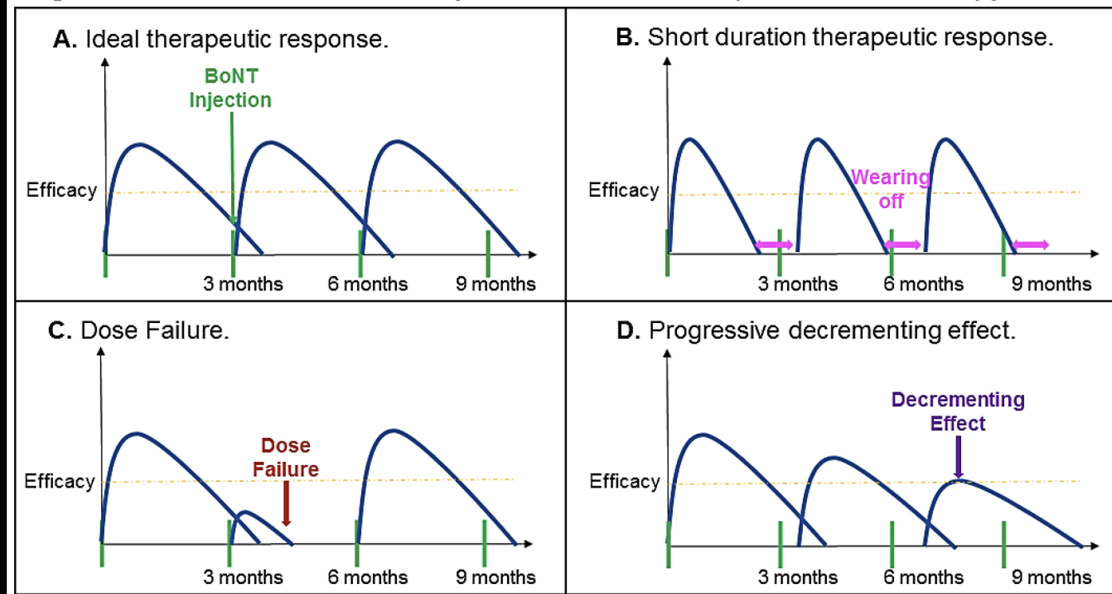
Brian D Berman ¹, Christopher L Groth,² Stefan H Sillau,¹ Sarah Pirio Richardson,³ Scott A Norris,⁴ Johanna Junker,^{5,6} Norbert Brüggemann ^{5,6}, Pinky Agarwal,⁷ Richard L Barbano,⁸ Alberto J Espay,⁹ Joaquin A Vizcarra,¹⁰ Christine Klein,⁶ Tobias Bäumer,⁶ Sebastian Loens,⁶ Stephen G Reich,¹¹ Marie Vidailhet,¹² Cecilia Bonnet,¹² Emmanuel Roze,¹² Hyder A Jinnah,¹³ Joel S Perlmuter¹⁴



Importance of Patient Experience



Figure 2. Fluctuations in severity over time and complications of therapy.



Importance of Objective Measures



Neurology 2016

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Objective, computerized video-based rating of blepharospasm severity

ABSTRACT

Objective: To compare clinical rating scales of blepharospasm severity with involuntary eye closures measured automatically from patient videos with contemporary facial expression software.

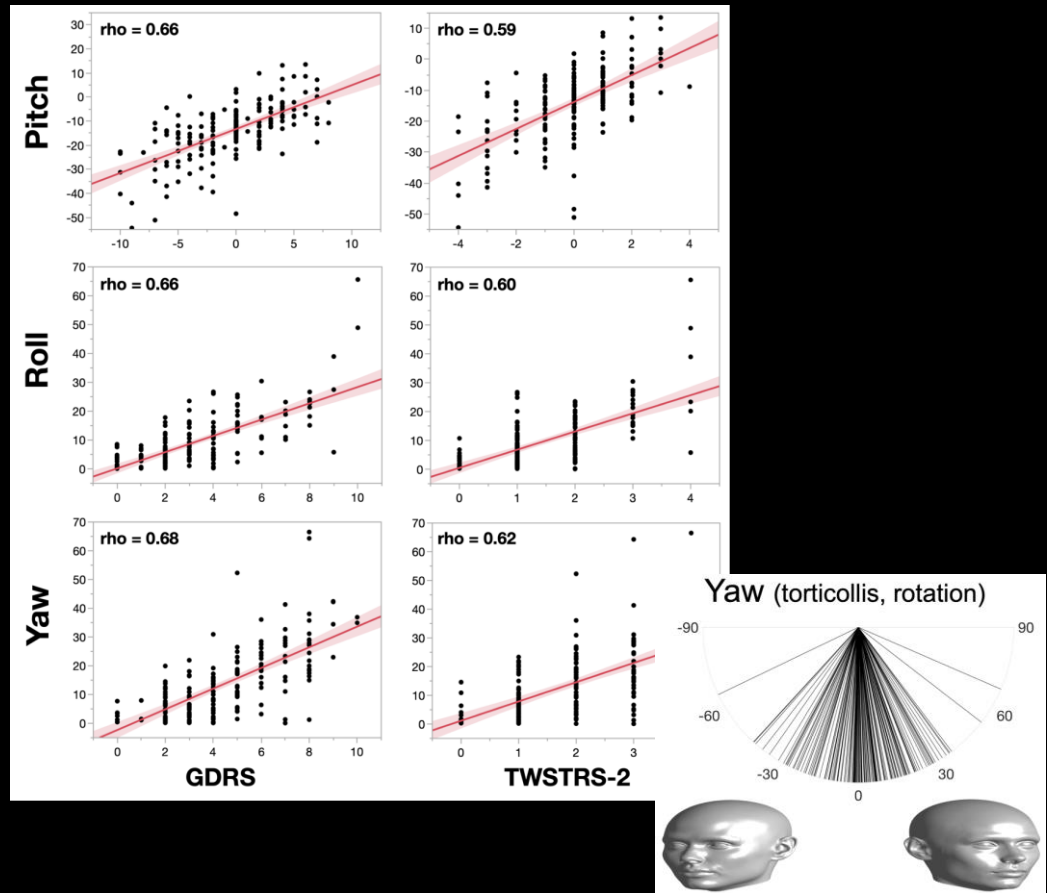
Methods: We evaluated video recordings of a standardized clinical examination from 50 patients with blepharospasm in the Dystonia Coalition's Natural History and Biorepository study. Eye closures were measured on a frame-by-frame basis with software known as the Computer Expression Recognition Toolbox (CERT). The proportion of eye closure time was compared with 3 commonly used clinical rating scales: the Burke-Fahn-Marsden Dystonia Rating Scale, Global Dystonia Rating Scale, and Jankovic Rating Scale.

Results: CERT was reliably able to find the face, and its eye closure measure was correlated with all of the clinical severity ratings (Spearman $\rho = 0.56, 0.52,$ and 0.56 for the Burke-Fahn-Marsden Dystonia Rating Scale, Global Dystonia Rating Scale, and Jankovic Rating Scale, respectively, all $p < 0.0001$).

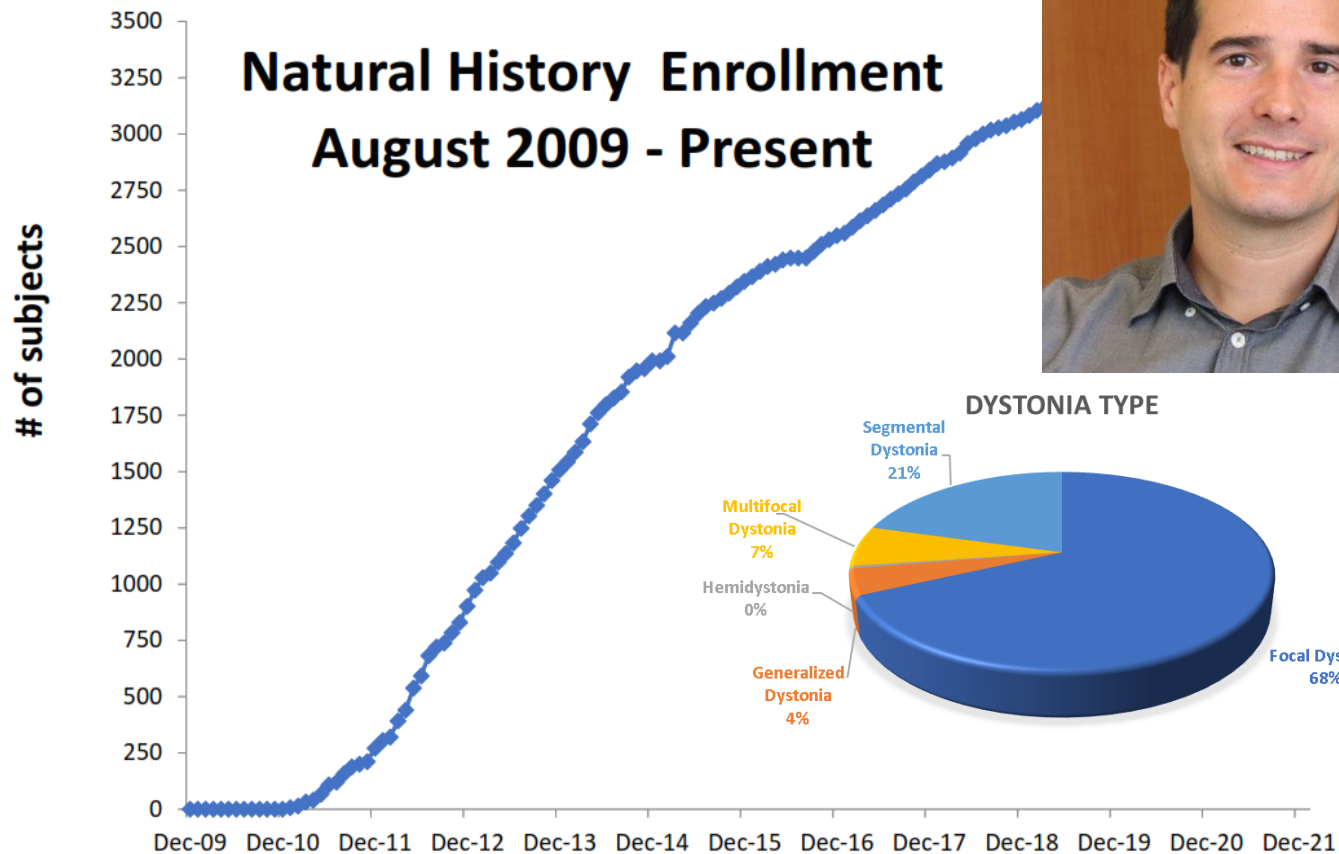
Conclusions: The results demonstrate that CERT has convergent validity with conventional clinical rating scales and can be used with video recordings to measure blepharospasm symptom severity automatically and objectively. Unlike EMG and kinematics, CERT requires only conventional video recordings and can therefore be more easily adopted for use in the clinic.

Neurology® 2016;87:2146–2153

Importance of Objective Measures



Importance of a Biobank



Experimental Therapeutics

EXPERT OPINION ON DRUG DISCOVERY
2019, VOL. 14, NO. 9, 893–900
<https://doi.org/10.1080/17460441.2019.1623785>



REVIEW



New approaches to discovering drugs that treat dystonia

Sarah Pirio Richardson^{a,b} and H. A. Jinnah^c

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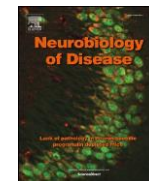
Neurobiology of Disease 130 (2019) 104526



Contents lists available at [ScienceDirect](https://www.sciencedirect.com)

Neurobiology of Disease

journal homepage: www.elsevier.com/locate/ynbdi



Review

The neurobiological basis for novel experimental therapeutics in dystonia

Anthony M. Downs^{a,1}, Kaitlyn M. Roman^{a,1}, Simone A. Campbell^a, Antonio Pisani^{b,c},
Ellen J. Hess^{a,d}, Paola Bonsi^{b,*}



Dystonia Coalition:

What have we done so far?

- Grown from 8 Sites to Many
 - 35 active or initiated recruiting centers*
 - 8 affiliate centers*
- Conducted Several Major Clinical Studies
 - all address bottlenecks in trial readiness*
 - all have international participation*
- Seeded Numerous Smaller Pilot Studies
 - 40 pilot projects*
 - 14 career awards*
- More than 100 publications
 - Brain, JAMA, J Neurosci, Mov Disord, Neurol*

Dystonia Coalition:

Data & sample sharing

- Main Research Projects
 - ~3200 subjects in Natural History Project*
 - ~200 subjects in CD Rating Scale Project*
 - ~200 subjects in LD Diagn & Rating Scale Project*
 - ~200 subjects in BSP Diagn & Rating Scale Project*
- What is available?
 - clinical data*
 - video recordings of exams*
 - DNA specimens (at Coriell)*
- How to request data or samples?
 - data/sample request form*
 - dystoniacoalition@emory.edu*
 - 56 requests made already*

Look Here for More Info:



The Dystonia Coalition: A Multicenter Network for Clinical and Translational Studies

REVIEW

published: 08 April 2021

doi: 10.3389/fneur.2021.660909

Gamze Kilic-Berkmen¹, Laura J. Wright², Joel S. Perlmutter³, Cynthia Comella⁴, Mark Hallett⁵, Jan Teller⁶, Sarah Pirio Richardson⁷, David A. Peterson⁸, Carlos Cruchaga⁹, Codrin Lungu¹⁰ and H. A. Jinnah^{1,11}*

- The need for trial readiness
- Summary of main projects
- Summary of pilots and career awards
- How to access data/materials
- How to get more involved

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Special thanks to some people who make everything happen!



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